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Researchers identify new gene mutation associated with ALS

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A research team led by investigators at the National Institute on Aging at the National Institutes of Health has discovered a new gene mutation associated with ALS, amyotrophic lateral sclerosis. The mutation is involved in RNA metabolism, which is part of the control mechanism determining protein synthesis. The findings appear in the March 30, 2014, issue of *Nature Neuroscience*.

ALS, often referred to as Lou Gehrig's disease, is a rapidly progressive, fatal neurological disorder that kills about 6,000 Americans each year. The disease attacks and kills nerve cells in the brain and spinal cord, and people with ALS lose strength and the ability to move their arms, legs, and body, and eventually, the ability to breathe without support. About 10 percent of people with ALS have a directly inherited form of the disease.

The discovery involves a mutation in the Matrin 3 gene, located on chromosome 5. The researchers applied exome sequencing to DNA samples from families in which several people had been diagnosed with ALS and identified the Matrin 3 mutation in a number of individuals. Further investigation revealed an interaction between the Matrin 3 protein and the TDP-43 protein, an RNA-binding protein whose mutation is known to cause ALS.

"The identification of this gene mutation gives us another target to explore in the pathogenesis of this disease," said senior author Bryan J. Traynor, M.D, Ph.D., of NIA's Laboratory of Neurogenetics. "It also

provides additional evidence that some disruption in RNA metabolism, an essential process within all cells, is involved in neuron death in ALS."

More information: "Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis" by Johnson, J.O., et al. *Nature Neuroscience*. Published online on March 30, 2014. DOI: 10.1038/nn.3688

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